



## Fuchs endothelial dystrophy

Fuchs endothelial dystrophy is a condition that causes vision problems. The first symptom of this condition is typically blurred vision in the morning that usually clears during the day. Over time, affected individuals lose the ability to see details (visual acuity). People with Fuchs endothelial dystrophy also become sensitive to bright lights.

Fuchs endothelial dystrophy specifically affects the front surface of the eye called the cornea. Deposits called guttae, which are detectable during an eye exam, form in the middle of the cornea and eventually spread. These guttae contribute to the loss of cells in the cornea, leading to vision problems. Tiny blisters may develop on the cornea, which can burst and cause eye pain.

The signs and symptoms of Fuchs endothelial dystrophy usually begin in a person's forties or fifties. A very rare early-onset variant of this condition starts to affect vision in a person's twenties.

### Frequency

The late-onset form of Fuchs endothelial dystrophy is a common condition, affecting approximately 4 percent of people over the age of 40. The early-onset variant of Fuchs endothelial dystrophy is rare, although the exact prevalence is unknown.

For reasons that are unclear, women are affected with Fuchs endothelial dystrophy somewhat more frequently than men.

### Genetic Changes

The genetics of Fuchs endothelial dystrophy are unclear. Researchers have identified regions of a few chromosomes and several genes that they think may play a role in the development of Fuchs endothelial dystrophy, but many of these associations need to be further tested.

Fuchs endothelial dystrophy affects a thin layer of cells that line the back of the cornea, called corneal endothelial cells. These cells regulate the amount of fluid inside the cornea. An appropriate fluid balance in the cornea is necessary for clear vision.

Fuchs endothelial dystrophy occurs when the endothelial cells die, and the cornea becomes swollen with too much fluid. Corneal endothelial cells continue to die over time, resulting in further vision problems. It is thought that mutations in genes that are active (expressed) primarily in corneal endothelial cells or surrounding tissue may lead to the death of corneal endothelial cells, resulting in Fuchs endothelial dystrophy.

Some cases of the early-onset variant of Fuchs endothelial dystrophy are caused by mutations in the COL8A2 gene. This gene provides instructions for making a protein

that is part of type VIII collagen. Type VIII collagen is largely found within the cornea, surrounding the endothelial cells. Specifically, type VIII collagen is a major component of a tissue at the back of the cornea, called Descemet's membrane. This membrane is a thin, sheet-like structure that separates and supports corneal endothelial cells. *COL8A2* gene mutations that cause the early-onset variant of Fuchs endothelial dystrophy lead to an abnormal Descemet's membrane, which causes the cells to die and leads to the vision problems in people with this condition.

Mutations in unidentified genes are also likely to cause the early-onset variant of Fuchs endothelial dystrophy. The genetic causes of the late-onset form of the disorder are unknown.

## Inheritance Pattern

In some cases, Fuchs endothelial dystrophy appears to be inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. When this condition is caused by a mutation in the *COL8A2* gene, it is inherited in an autosomal dominant pattern. In addition, an autosomal dominant inheritance pattern is apparent in some situations in which the condition is caused by alterations in an unknown gene.

In many families, the inheritance pattern is unknown.

Some cases result from new mutations in a gene and occur in people with no history of the disorder in their family.

## Other Names for This Condition

- Fuchs atrophy
- Fuchs corneal dystrophy
- Fuchs dystrophy
- Fuchs endothelial corneal dystrophy
- Fuchs' endothelial dystrophy

## Diagnosis & Management

### Genetic Testing

- Genetic Testing Registry: Corneal dystrophy, Fuchs endothelial 1  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1850959/>
- Genetic Testing Registry: Corneal dystrophy, Fuchs endothelial, 2  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C1857800/>
- Genetic Testing Registry: Corneal dystrophy, Fuchs endothelial, 3  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2750451/>

- Genetic Testing Registry: Corneal dystrophy, Fuchs endothelial, 4  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2750450/>
- Genetic Testing Registry: Corneal dystrophy, Fuchs endothelial, 5  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2750449/>
- Genetic Testing Registry: Corneal dystrophy, Fuchs endothelial, 6  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2750448/>
- Genetic Testing Registry: Corneal dystrophy, Fuchs endothelial, 7  
<https://www.ncbi.nlm.nih.gov/gtr/conditions/C2750447/>

#### Other Diagnosis and Management Resources

- Duke Health: Corneal Disease  
<https://www.dukehealth.org/treatments/eye-care/cornea-disease>
- MedlinePlus Encyclopedia: Fuchs Dystrophy  
<https://medlineplus.gov/ency/article/007295.htm>

#### General Information from MedlinePlus

- Diagnostic Tests  
<https://medlineplus.gov/diagnostictests.html>
- Drug Therapy  
<https://medlineplus.gov/drugtherapy.html>
- Genetic Counseling  
<https://medlineplus.gov/geneticcounseling.html>
- Palliative Care  
<https://medlineplus.gov/palliativecare.html>
- Surgery and Rehabilitation  
<https://medlineplus.gov/surgeryandrehabilitation.html>

#### **Additional Information & Resources**

##### MedlinePlus

- Encyclopedia: Fuchs Dystrophy  
<https://medlineplus.gov/ency/article/007295.htm>
- Health Topic: Corneal Disorders  
<https://medlineplus.gov/cornealdisorders.html>
- Health Topic: Vision Impairment and Blindness  
<https://medlineplus.gov/visionimpairmentandblindness.html>

## Genetic and Rare Diseases Information Center

- Fuchs endothelial corneal dystrophy  
<https://rarediseases.info.nih.gov/diseases/10018/fuchs-endothelial-corneal-dystrophy>

## Additional NIH Resources

- National Eye Institute: Cornea and Corneal Disease  
<https://nei.nih.gov/health/cornealdisease/>
- National Eye Institute: Low Vision  
<https://nei.nih.gov/health/lowvision/>

## Educational Resources

- Cleveland Clinic: Corneal Conditions  
<http://my.clevelandclinic.org/health/articles/corneal-conditions>
- Digital Reference of Ophthalmology  
<http://dro.hs.columbia.edu/fuchs2.htm>
- Disease InfoSearch: Corneal Dystrophy Fuchs Endothelial 1  
<http://www.diseaseinfosearch.org/Corneal+Dystrophy+Fuchs+Endothelial+1/1908>
- MalaCards: fuchs' endothelial dystrophy  
[http://www.malacards.org/card/fuchs\\_endothelial\\_dystrophy](http://www.malacards.org/card/fuchs_endothelial_dystrophy)
- Orphanet: Fuchs endothelial corneal dystrophy  
[http://www.orpha.net/consor/cgi-bin/OC\\_Exp.php?Lng=EN&Expert=98974](http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=98974)
- Wilmer Eye Institute at Johns Hopkins: What is Fuchs Endothelial Corneal Dystrophy?  
<http://www.hopkinsmedicine.org/wilmer/conditions/Fuchs/about/>

## Patient Support and Advocacy Resources

- American Foundation for the Blind  
<http://www.afb.org/>
- Cornea Research Foundation of America  
<http://www.cornea.org/>
- Corneal Dystrophy Foundation  
<https://www.cornealdystrophyfoundation.org/>
- National Organization for Rare Disorders (NORD): Corneal Dystrophies  
<https://rarediseases.org/rare-diseases/corneal-dystrophies/>

- Prevent Blindness America  
<http://www.preventblindness.org/>
- The Foundation Fighting Blindness  
<http://ffb.ca/>

#### ClinicalTrials.gov

- ClinicalTrials.gov  
<https://clinicaltrials.gov/ct2/results?cond=%22Fuchs+endothelial+dystrophy%22+OR+%22Corneal+Granular+Dystrophies%22+OR+%22Hereditary+Corneal+Dystrophies%22+OR+%22Corneal+Macular+Dystrophy%22>

#### Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28Fuchs'+Endothelial+Dystrophy%5BMAJR%5D%29+AND+%28%28Fuchs+endothelial+dystrophy%5BTIAB%5D%29+OR+%28Fuchs+corneal+dystrophy%5BTIAB%5D%29+OR+%28Fuchs+endothelial+corneal+dystrophy%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

#### OMIM

- CORNEAL DYSTROPHY, FUCHS ENDOTHELIAL, 1  
<http://omim.org/entry/136800>
- CORNEAL DYSTROPHY, FUCHS ENDOTHELIAL, 2  
<http://omim.org/entry/610158>
- CORNEAL DYSTROPHY, FUCHS ENDOTHELIAL, 3  
<http://omim.org/entry/613267>
- CORNEAL DYSTROPHY, FUCHS ENDOTHELIAL, 4  
<http://omim.org/entry/613268>
- CORNEAL DYSTROPHY, FUCHS ENDOTHELIAL, 5  
<http://omim.org/entry/613269>
- CORNEAL DYSTROPHY, FUCHS ENDOTHELIAL, 6  
<http://omim.org/entry/613270>
- CORNEAL DYSTROPHY, FUCHS ENDOTHELIAL, 7  
<http://omim.org/entry/613271>

## Sources for This Summary

- Afshari NA, Li YJ, Pericak-Vance MA, Gregory S, Klintworth GK. Genome-wide linkage scan in fuchs endothelial corneal dystrophy. *Invest Ophthalmol Vis Sci.* 2009 Mar;50(3):1093-7. doi: 10.1167/iovs.08-1839. Epub 2008 May 23.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/18502986>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2712757/>
- Baratz KH, Tosakulwong N, Ryu E, Brown WL, Branham K, Chen W, Tran KD, Schmid-Kubista KE, Heckenlively JR, Swaroop A, Abecasis G, Bailey KR, Edwards AO. E2-2 protein and Fuchs's corneal dystrophy. *N Engl J Med.* 2010 Sep 9;363(11):1016-24. doi: 10.1056/NEJMoa1007064. Epub 2010 Aug 25.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/20825314>
- Biswas S, Munier FL, Yardley J, Hart-Holden N, Perveen R, Cousin P, Sutphin JE, Noble B, Batterbury M, Kiely C, Hackett A, Bonshek R, Ridgway A, McLeod D, Sheffield VC, Stone EM, Schorderet DF, Black GC. Missense mutations in COL8A2, the gene encoding the alpha2 chain of type VIII collagen, cause two forms of corneal endothelial dystrophy. *Hum Mol Genet.* 2001 Oct 1; 10(21):2415-23.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/11689488>
- Eghrari AO, Gottsch JD. Fuchs' corneal dystrophy. *Expert Rev Ophthalmol.* 2010 Apr;5(2):147-159.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/20625449>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2897712/>
- Gottsch JD, Sundin OH, Liu SH, Jun AS, Broman KW, Stark WJ, Vito EC, Narang AK, Thompson JM, Magovern M. Inheritance of a novel COL8A2 mutation defines a distinct early-onset subtype of fuchs corneal dystrophy. *Invest Ophthalmol Vis Sci.* 2005 Jun;46(6):1934-9.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/15914606>
- Riazuddin SA, Vithana EN, Seet LF, Liu Y, Al-Saif A, Koh LW, Heng YM, Aung T, Meadows DN, Eghrari AO, Gottsch JD, Katsanis N. Missense mutations in the sodium borate cotransporter SLC4A11 cause late-onset Fuchs corneal dystrophy. *Hum Mutat.* 2010 Nov;31(11):1261-8. doi: 10.1002/humu.21356. Epub 2010 Oct 14.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/20848555>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2970683/>
- Riazuddin SA, Zaghloul NA, Al-Saif A, Davey L, Diplas BH, Meadows DN, Eghrari AO, Minear MA, Li YJ, Klintworth GK, Afshari N, Gregory SG, Gottsch JD, Katsanis N. Missense mutations in TCF8 cause late-onset Fuchs corneal dystrophy and interact with FCD4 on chromosome 9p. *Am J Hum Genet.* 2010 Jan;86(1):45-53. doi: 10.1016/j.ajhg.2009.12.001. Epub 2009 Dec 31.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/20036349>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2801746/>
- Vithana EN, Morgan PE, Ramprasad V, Tan DT, Yong VH, Venkataraman D, Venkatraman A, Yam GH, Nagasamy S, Law RW, Rajagopal R, Pang CP, Kumaramanickevel G, Casey JR, Aung T. SLC4A11 mutations in Fuchs endothelial corneal dystrophy. *Hum Mol Genet.* 2008 Mar 1;17(5): 656-66. Epub 2007 Nov 16.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/18024964>
- Wright AF, Dhillon B. Major progress in Fuchs's corneal dystrophy. *N Engl J Med.* 2010 Sep 9; 363(11):1072-5. doi: 10.1056/NEJMe1007495. Epub 2010 Aug 25.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/20825321>

---

Reprinted from Genetics Home Reference:  
<https://ghr.nlm.nih.gov/condition/fuchs-endothelial-dystrophy>

Reviewed: June 2011  
Published: March 21, 2017

Lister Hill National Center for Biomedical Communications  
U.S. National Library of Medicine  
National Institutes of Health  
Department of Health & Human Services